Aberrant alternative splicing yields a secreted FGFR2 IIIa TM disease variant

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Introduction

Reactome is open-source, open access, manually curated and peer-reviewed pathway database. Pathway annotations are authored by expert biologists, in collaboration with Reactome editorial staff and cross-referenced to many bioinformatics databases. A system of evidence tracking ensures that all assertions are backed up by the primary literature. Reactome is used by clinicians, geneticists, genomics researchers, and molecular biologists to interpret the results of high-throughput experimental studies, by bioinformaticians seeking to develop novel algorithms for mining knowledge from genomic studies, and by systems biologists building predictive models of normal and disease variant pathways.

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Literature references

Fabregat, A., Sidiropoulos, K., Viteri, G., Forner, O., Marin-Garcia, P., Arnau, V. et al. (2017). Reactome pathway analysis: a high-performance in-memory approach. *BMC bioinformatics, 18*, 142. ↗

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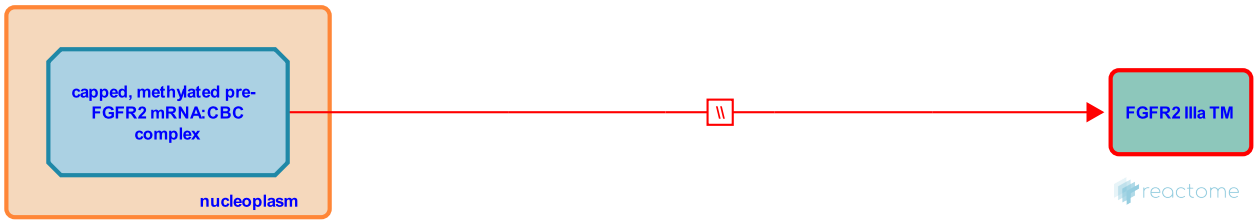
Stable identifier: R-HSA-8851710

Type: omitted

Compartments: extracellular region

Diseases: acrocephalosyndactylia

Inferred from: Aberrant alternative splicing generates a soluble disease variant FGFR2 IIIa TM (Mus musculus)



A secreted truncated form of FGFR2 known as IIIa TM is produced and stable in a mouse model of Apert Syndrome. FGFR2 IIIa TM is formed from aberrant splicing of FGFR2 exon 7 (IIIa) into exon 10 (containing the transmembrane domain). In WT cells, this transcript is degraded by nonsense-mediated decay, but persists in the disease model by an unknown mechanism. FGFR IIIa TM modulates the binding of FGF1 to FGFR2 in vitro and negatively regulates FGFR2 signaling in vitro and in vivo (Wheldon et al, 2011).

Literature references

Hajihosseini, MK., Khodabukus, N., Heath, JK., Smith, TG., Patey, SJ., Wheldon, LM. (2011). Identification and characterization of an inhibitory fibroblast growth factor receptor 2 (FGFR2) molecule, up-regulated in an Apert Syndrome mouse model. *Biochem. J., 436*, 71-81. ↗

Editions

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